



Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the gene(s) analyzed. NGS, multiplex ligation-dependent probe amplification (MLPA), and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the gene(s) analyzed. PCR and gel electrophoresis is performed to test for the presence of the 10 megabase inversion of coding exons 1–7 of the *MSH2* gene. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript^a
<i>APC</i>	NM_000038.6
<i>ATM</i>	NM_000051.3
<i>AXIN2</i>	NM_004655.4
<i>BARD1</i>	NM_000465.4
<i>BMPR1A</i>	NM_004329.2
<i>BRCA1</i>	NM_007294.4
<i>BRCA2</i>	NM_000059.3
<i>BRIP1</i>	NM_032043.3
<i>CDH1</i>	NM_004360.5
<i>CDK4</i>	NM_000075.4
<i>CDKN2A</i>	NM_000077.4
<i>CHEK2</i>	NM_007194.4
<i>DICER1</i>	NM_177438.2
<i>EPCAM^c</i>	NM_002354.3
<i>GREM1^d</i>	NM_013372.7
<i>HOXB13</i>	NM_006361.5
<i>MEN1</i>	NM_130799.2
<i>MLH1</i>	NM_000249.3

Gene	Reference Transcript^a
<i>MSH2</i>	NM_000251.3
<i>MSH3</i>	NM_002439.5
<i>MSH6</i>	NM_000179.2
<i>MUTYH</i>	NM_001128425.1
<i>NBN^b</i>	NM_002485.4
<i>NF1</i>	NM_000267.3
<i>NTHL1</i>	NM_002528.7
<i>PALB2</i>	NM_024675.4
<i>PMS2</i>	NM_000535.7
<i>POLD1</i>	NM_002691.4
<i>POLE</i>	NM_006231.4
<i>PTEN</i>	NM_000314.8
<i>RAD51C</i>	NM_058216.3
<i>RAD51D</i>	NM_002878.3
<i>RET</i>	NM_020975.6
<i>SMAD4</i>	NM_005359.6
<i>STK11</i>	NM_000455.5
<i>TP53</i>	NM_000546.5

^a Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

^b There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

^c This gene is analyzed for the presence of copy number variants only. No sequencing analysis is performed.

^d The *GREM1* gene is analyzed for the presence of copy number gains involving the upstream enhancer region only. No other sequencing or copy number variant analyses are performed.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.